Central Connecticut Cystic Fibrosis Center

Testimony of Craig Lapin, MD, Director, Central Connecticut Cystic Fibrosis Center and Associate Professor of Pediatrics, University of Connecticut Health Center to the Public Health Committee regarding *House Bill 6263, An Act Requiring The Administration Of A Screening Test For Cystic Fibrosis To Newborn Infants.*

February 6, 2009

Senator Harris, Representative Ritter, Members of the Public Health Committee. Thank you for the opportunity to testify in support of *House Bill #6263, An Act Requiring The Administration of a Screening Test for Cystic Fibrosis to Newborn Infants.* My name is Dr. Craig Lapin and I am Director of the Central Connecticut Cystic Fibrosis Center and Associate Professor of Pediatrics at the University of Connecticut Health Center.

Last Thursday we had our first infant identified with CF by NBS for 2009; they had the diagnosis confirmed on Friday; the family had a 2 hour meeting with the CF center on three days ago, and the baby was seen Wednesday. That is the power and benefit of the CF NBS program. If this infant had not been born at a hospital that does screening, we do not know when and how sick he would have been when finally diagnosed.

You will be hearing the personal impact that a delayed diagnosis has for families with cystic fibrosis. I would like to present the medical case to add cystic fibrosis to mandated newborn screening (NBS). CF is the most common lethally inherited disease in caucasians, although it also affects other ethnicities as well. CF occurs in 1 in 3000 newborns. It occurs 3 times more frequently than Phenylketonuria and 50 times more than Maple Syrup Urine Disease. In past two years screening for the whole of Connecticut there have been six cases of phenylketonuria, 5 medium-chain acyl-dehyrogenase deficiency, one long-chain acyl-dehyrogenase deficiency, no maple syrup urine disease; these are all amongst the screened diseases here in Connecticut. For the past two years, screening at UCONN/CCMC alone has identified seven cases of CF. This is not meant as an argument not to screen for other diseases, just that from a public health standpoint, cystic fibrosis occurs as often as or more frequently than most of the diseases for which we currently screen.

CF causes multiple problems but primary are respiratory and nutritional. There is extensive medical research that shows early diagnosis makes a significant difference in the health outcomes (and therefore lives) of patients with CF. In the short term – for infants diagnosed at less than 1 year of age because of CF symptoms (i.e. not by NBS) 33% were grossly malnourished compared with 11% diagnosed by NBS. Significant CF infections were found twice as frequently (29% not NBS vs 15% NBS), and patients were hospitalized three times as much (64% not NBS vs 22% NBS). Of even greater concern,

5% of patient diagnosed with CF over a 3 year period had life-threatening malnutrition, compared with none by NBS.

In the long term over years, the national CF registry database shows that as people with CF grow older, for every age group, those diagnosed symptomatically are always at least twice as likely to be malnourished compared with those diagnosed by NBS. Those diagnosed with CF by NBS are statistically less likely to be stunted, or to have the special CF infections that lead to more rapid pulmonary function decline and therefore decreased quality of life. By the second decade of life, people diagnosed by NBS are less likely to require hospitalization (thus significantly decreasing cost of care).

Solid research and multiple studies, document that delayed diagnosis of CF and malnutrition in this disease leads to failure to thrive, increased infections, a more rapid decline in lung function, subsequent decreased quality of life, and decreased life span. In other words, people die earlier. Waiting until patients have symptoms of CF is associated with higher complications rates and morbidity compared to diagnosis by NBS. Dr. Collins recently published data from Connecticut showing that patients diagnosed by NBS maintain significantly better pulmonary function and nutrition.

The Center for Disease Control (CDC) has determined that screening for CF is justified. Screening for CF involves a two-stage process performed on the single heelstick done at birth. Currently, the University of Connecticut Health Center (UCHC) and Yale New Haven Hospital (Yale) perform the tests using blood samples provided by the 20 of the 30 birthing hospitals in Connecticut. We estimate there are approximately 13,000 Connecticut babies unscreened each year, or over 200,000 children since 1993 when the voluntary CF NBS program began. After a positive screen the family's primary care provider is notified by phone, fax, and registered letter recommending the infant be sent to one of the two Cystic Fibrosis treatment centers in Connecticut (UCHC/Connecticut Children's or Yale) for a definitive sweat test following a positive Stage 2 test.

Our CF center has been part of the voluntary newborn screening program that has been extremely efficient and supportive of families for sixteen years, screening approximately 26,000 infants a year. Yale screens approximately 8,000 patients a year. Please, for the sakes of the infants, children, and adults with CF, mandate newborn screening for cystic fibrosis that extends the current program to all. Thank you.

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The Case for Newborn Screening for Cystic Fibrosis

The Disease

- Cystic Fibrosis (CF) is a chronically debilitating genetic disease that affects the respiratory, gastrointestinal and reproductive systems.
- CF occurs in approximately one of every 3,500 live births and about 1,000 new cases of CF are diagnosed each year.

Diagnosis

- The major symptoms of CF are not unique to the disease.
- According to the CDC, half of all individuals in the United States with CF were diagnosed after six months of age.
- Universal newborn screening would prevent delayed diagnoses.

Newborn Screening

- Voluntary newborn screening for CF began in Connecticut in 1993 – one of the first states in the country to do so. If not mandated, by the end of 2009 Connecticut will be the only state in the country that does NOT screen all its newborns.
- 20 of Connecticut's 30 birthing hospitals participate in the voluntary program.
  - ◊ As a result, about 2/3 of Connecticut newborns are tested.
  - ◊ Of the 10 hospitals that do not participate, Danbury and other Fairfield area hospitals account for most of the births.
- The cost of the blood test for CF (called the immunoreactive trypsinogen test or IRT) is $15-$20 per child.
- An HRSA-commissioned report released on March 8, 2005, calls for the standardization of newborn screening tests throughout the country; CF is on the report's list of 29 recommended tests. The report is available at http://genes-r-us.uthscsa.edu.

The Impact of Early Detection

- Early diagnosis leads to immediate intervention with specialized therapies that include pancreatic enzymes to aid digestion and a high-calorie, high-fat diet.
- Immediate interventions result in:
  - ◊ Improved height, weight and cognitive function
  - ◊ Decreased risk of life-threatening malnutrition
  - ◊ The maintenance of respiratory function
  - ◊ Increased life expectancy, and
  - ◊ Reduced hospitalizations